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FULL ESTIMATED COST                                         2.31       2.31

FILE 'MEDLINE' ENTERED AT 11:26:44 ON 22 JUL 2002

FILE 'BIOSIS' ENTERED AT 11:26:44 ON 22 JUL 2002
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FILE 'CAPLUS' ENTERED AT 11:26:44 ON 22 JUL 2002
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=> s single (w) (nucleotide or base) (w) exten?
L1          147 SINGLE (W) (NUCLEOTIDE OR BASE) (W) EXTEN?

=> s l1 and (rolling (w) circle)
L2            3 L1 AND (ROLLING (W) CIRCLE)

=> d 1-3 ti

L2   ANSWER 1 OF 3     MEDLINE
TI   Enabling large-scale pharmacogenetic studies by high-throughput mutation
     detection and genotyping technologies.

L2   ANSWER 2 OF 3  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI   Enabling large-scale pharmacogenetic studies by high-throughput mutation
     detection and genotyping technologies.

L2   ANSWER 3 OF 3  CAPLUS  COPYRIGHT 2002 ACS
TI   Enabling large-scale pharmacogenetic studies by high-throughput mutation
     detection and genotyping technologies


=> d bib ab

L2   ANSWER 1 OF 3     MEDLINE
AN   2001156126     MEDLINE
DN   21098045   PubMed ID: 11159763
TI   Enabling large-scale pharmacogenetic studies by high-throughput mutation
     detection and genotyping technologies.
AU   Shi M M
CS   Department of Applied Genomics, Genometrix Inc., The Woodlands, TX 77381,
     USA.. mshi@genometrix.com
SO   CLINICAL CHEMISTRY, (2001 Feb) 47 (2) 164-72.  Ref: 40
     Journal code: 9421549. ISSN: 0009-9147.
CY   United States
DT   Journal; Article; (JOURNAL ARTICLE)
     General Review; (REVIEW)
     (REVIEW, TUTORIAL)
LA   English
FS   Priority Journals
EM   200103
ED   Entered STN: 20010404
     Last Updated on STN: 20010404
     Entered Medline: 20010322
AB   BACKGROUND: Pharmacogenetics is a scientific discipline that examines the
     genetic basis for individual variations in response to therapeutics.
     Pharmacogenetics promises to develop individualized medicines tailored to
     patients' genotypes. However, identifying and genotyping a vast number of
```

genetic polymorphisms in large populations also pose a great challenge. APPROACH: This article reviews the recent technology development in mutation detection and genotyping with a focus on genotyping of single nucleotide polymorphisms (SNPs). CONTENT: Novel mutations/polymorphisms are commonly identified by conformation-based mutation screening and direct high-throughput heterozygote sequencing. With a large amount of public sequence information available, in silico SNP mapping has also emerged as a cost-efficient way for new polymorphism identification. Gel electrophoresis-based genotyping methods for known polymorphisms include PCR coupled with restriction fragment length polymorphism analysis, multiplex PCR, oligonucleotide ligation assay, and minisequencing. Fluorescent dye-based genotyping technologies are emerging as high-throughput genotyping platforms, including oligonucleotide ligation assay, pyrosequencing, **single-base extension** with fluorescence detection, homogeneous solution hybridization such as TaqMan, and molecular beacon genotyping. **Rolling circle** amplification and Invader assays are able to genotype directly from genomic DNA without PCR amplification. DNA chip-based microarray and mass spectrometry genotyping technologies are the latest development in the genotyping arena. SUMMARY: Large-scale genotyping is crucial to the identification of the genetic make-ups that underlie the onset of diseases and individual variations in drug responses. Enabling technologies to identify genetic polymorphisms rapidly, accurately, and cost effectively will dramatically impact future drug and development processes.

```
=> s l1 and lizardi?/au
L3            0 L1 AND LIZARDI?/AU

=> s l1 and amplif?
L4           48 L1 AND AMPLIF?

=> dup rem l4
PROCESSING COMPLETED FOR L4
L5           32 DUP REM L4 (16 DUPLICATES REMOVED)

=> d 1-32 ti

L5   ANSWER 1 OF 32  CAPLUS  COPYRIGHT 2002 ACS
TI   Method for determining alleles with polymorphic sites by hybridization,
     single base extension and ligation

L5   ANSWER 2 OF 32  CAPLUS  COPYRIGHT 2002 ACS
TI   Determining the degree of methylation of defined cytosines in genomic DNA
     in the sequence context 5'-CpG-3' by bisulfite modification

L5   ANSWER 3 OF 32  CAPLUS  COPYRIGHT 2002 ACS
TI   Devices and methods to form a randomly ordered array of magnetic beads and
     uses thereof in high-throughput genotyping

L5   ANSWER 4 OF 32     MEDLINE                              DUPLICATE 1
TI   Identification and minisequencing-based discrimination of SHV
     beta-lactamases in nosocomial infection-associated Klebsiella pneumoniae
     in Brisbane, Australia.

L5   ANSWER 5 OF 32  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.DUPLICATE
     2
TI   Molecular tagging of the Ms locus in onion.

L5   ANSWER 6 OF 32     MEDLINE                              DUPLICATE 3
TI   Accuracy of genotyping for single nucleotide polymorphisms by a
     microarray-based single nucleotide polymorphism typing method involving
```

hybridization of short allele-specific oligonucleotides.

L5 ANSWER 7 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Methods and compositions relating to electrical detection of nucleic acid hybridization or peptide binding preferably using AC impedance

L5 ANSWER 8 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Three-dimensional microarray system for parallel genotyping of single nucleotide polymorphisms by PCR

L5 ANSWER 9 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Generic SBE-FRET protocol

L5 ANSWER 10 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Single tube SNP genotyping using mini-extension coupled with IR-labeled AcycloTerminatorsTM.

L5 ANSWER 11 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Genotyping of two mutations in the HFE gene using **single-base extension** and high-performance liquid chromatography

L5 ANSWER 12 OF 32 MEDLINE DUPLICATE 4
TI High-performance liquid chromatography multiplex detection of two single nucleotide mutations associated with hereditary hemochromatosis.

L5 ANSWER 13 OF 32 MEDLINE DUPLICATE 5
TI Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies.

L5 ANSWER 14 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Parallel primer extension approach to nucleic acid sequence analysis.

L5 ANSWER 15 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Primer extension on a microarray of gel-immobilized primers

L5 ANSWER 16 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Method for the analysis of single nucleotide polymorphisms by primer extension techniques in restriction fragments generated using AFLP

L5 ANSWER 17 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI Method for the analysis of AFLP reaction mixtures using primer extension techniques to detect specific restriction fragments

L5 ANSWER 18 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Simple two-color array-based approach for mutation detection.

L5 ANSWER 19 OF 32 MEDLINE DUPLICATE 6
TI Parallel genotyping of human SNPs using generic high-density oligonucleotide tag arrays.

L5 ANSWER 20 OF 32 MEDLINE DUPLICATE 7
TI Detection of single nucleotide polymorphisms of the human mu opioid receptor gene by hybridization or **single nucleotide extension** on custom oligonucleotide gelpad microchips: potential in studies of addiction.

L5 ANSWER 21 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies.

L5 ANSWER 22 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.

TI Mutation detection by **single nucleotide extension**.

L5 ANSWER 23 OF 32 MEDLINE DUPLICATE 8
TI Quantitative analysis of human DNA sequences by PCR and solid-phase minisequencing.

L5 ANSWER 24 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Multiple, time-spaced injections onto the MegaBACETM for high-throughput SNP genotyping.

L5 ANSWER 25 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Genotyping of HPA-1 (human platelet antigen 1) by mini-sequencing.

L5 ANSWER 26 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Genotyping using arrayed **single-base extension** .

L5 ANSWER 27 OF 32 CAPLUS COPYRIGHT 2002 ACS
TI **Amplification** and other enzymic reactions performed on nucleic acid arrays

L5 ANSWER 28 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
TI Single nucleotide polymorphism determination using primer extension and time-of-flight mass spectrometry.

L5 ANSWER 29 OF 32 MEDLINE DUPLICATE 9
TI A sensitive new method for rapid detection of abnormal methylation patterns in global DNA and within CpG islands.

L5 ANSWER 30 OF 32 MEDLINE
TI Direct sequencing of RAPD fragments using 3'-extended oligonucleotide primers and dye terminator cycle-sequencing.

L5 ANSWER 31 OF 32 MEDLINE
TI Polymorphism analysis and gene detection by minisequencing on an array of gel-immobilized primers.

L5 ANSWER 32 OF 32 MEDLINE DUPLICATE 10
TI Multiplex, fluorescent, solid-phase minisequencing for efficient screening of DNA sequence variation.

=> d 22 bib ab

L5 ANSWER 22 OF 32 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2000:360971 BIOSIS
DN PREV200000360971
TI Mutation detection by **single nucleotide extension**.
AU Moutereau, Stephane (1); Johnson, M. D.; Sakazume, T.; Rappaport, E.; Santacroce, R.; Graves, D.; Su, H.-J.; Delgrosso, K.; McKenzie, S.; Dong, P.; Surrey, S.; Fortina, P.
CS (1) Hopital Henri Mondor, Creteil France
SO European Journal of Human Genetics, (June, 2000) Vol. 8 , No. Supplement 1, pp. 129. print.
Meeting Info.: European Human Genetics Conference 2000 Amsterdam, Netherlands May 27-February 30, 2000 European Society of Human Genetics . ISSN: 1018-4813.
DT Conference
LA English
SL English

=> d his

(FILE 'HOME' ENTERED AT 11:20:22 ON 22 JUL 2002)

FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 11:26:44 ON 22 JUL 2002
L1 147 S SINGLE (W) (NUCLEOTIDE OR BASE) (W) EXTEN?
L2 3 S L1 AND (ROLLING (W) CIRCLE)
L3 0 S L1 AND LIZARDI?/AU
L4 48 S L1 AND AMPLIF?
L5 32 DUP REM L4 (16 DUPLICATES REMOVED)

=> s l1 and coupl?
L6 14 L1 AND COUPL?

=> dup rem l6
PROCESSING COMPLETED FOR L6
L7 11 DUP REM L6 (3 DUPLICATES REMOVED)

=> d 1-11 bib ab

L7 ANSWER 1 OF 11 CAPLUS COPYRIGHT 2002 ACS
AN 2002:172161 CAPLUS
DN 136:227897
TI Method for determining alleles with polymorphic sites by hybridization, **single base extension** and ligation
IN Liu, Xiangjun
PA Haplogen, LLC, USA
SO PCT Int. Appl., 38 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN.CNT 1

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2002018659	A2	20020307	WO 2001-US41956	20010830

W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PH, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG

PRAI US 2000-228994P P 20000830
AB The invention is relates to detg. alleles by identifying one or more polymorphic sites in a gene. The present invention provides methods and kits for sepg. and identifying alleles, and thereby the haplotype, in genomic DNA samples. The method generally involves hybridizing primers specific to polymorphic sites within the alleles to the DNA sample, elongating the primers by one or more nucleic acids, sepg. the elongated primers and identifying the alleles utilizing the elongated primer. The method also allows for a ligation of two primers, their sepn. and subsequent use in identifying the targeted allele. The method further provides that another primer can be used as a blocking site for elongation of the first primer such that a stretch of DNA that includes a polymorphic site is replicated and identified. The unextended or extended primers can be labeled so that the primer can be easily sepd. and/or identified.

L7 ANSWER 2 OF 11 CAPLUS COPYRIGHT 2002 ACS

AN 2002:488155 CAPLUS
DN 137:43871
TI Devices and methods to form a randomly ordered array of magnetic beads and uses thereof in high-throughput genotyping
IN Jain, Maneesh; White, Robert L.; Roberts, Lester A.
PA USA
SO U.S. Pat. Appl. Publ., 41 pp., which
CODEN: USXXCO
DT Patent
LA English
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	US 2002081714	A1	20020627	US 2001-923752	20010807
PRAI	US 2000-202357P	P	20000505		
	US 2000-223125P	P	20000807		

AB The invention includes devices and methods for forming random arrays of magnetic particles, arrays formed using these devices and methods, and to methods of using the arrays. The invention provides an assembly (chip) with magnetic domains that produce localized magnetic fields capable of immobilizing magnetic particles such as com. available magnetic beads. Probe or sensor mols. can be **coupled** to the beads, which are then dispersed on the assembly, forming a random order array. The arrays can be used for analyzing samples, targets, and/or the interaction between samples and targets. The invention finds particular use in processes such as high-throughput genotyping and other nucleic acid hybridization-based assays. The invention offers a no. of significant advantages in comparison with traditional DNA arrays in which probes are bound to a substrate.

L7 ANSWER 3 OF 11 CAPLUS COPYRIGHT 2002 ACS
AN 2002:446392 CAPLUS
DN 137:42259
TI A **single base extension** technique for the analysis of known mutations utilizing capillary gel electrophoresis with electrochemical detection
AU Brazill, Sara A.; Kuhr, Werner G.
CS Department of Chemistry, University of California, Riverside, CA, 92521, USA
SO Analytical Chemistry (2002), 74(14), 3421-3428
CODEN: ANCHAM; ISSN: 0003-2700
PB American Chemical Society
DT Journal
LA English
AB A novel single nucleotide polymorphism (SNP) detection system is described in which the accuracy of DNA polymerase and advantages of electrochem. detection are demonstrated. A model SNP system is presented to illustrate the potential advantages in **coupling** the **single base extension** (SBE) technique to capillary gel electrophoresis (CGE) with electrochem. detection. An electrochem. labeled primer, with a ferrocene acetate covalently attached to its 5' end, is used in the extension reaction. When the Watson-Crick complementary ddNTP is added to the SBE reaction, the primer is extended by a single nucleotide. The reaction mixt. is subsequently sepd. by CGE, and the ferrocene-tagged fragments are detected at the sepn. anode with sinusoidal voltammetry. This work demonstrates the first single base resoln. sepn. of DNA **coupled** with electrochem. detection. The unextended primer (20-mer) and the 21-mer extension product are sepd. with a resoln. of 0.8.
RE.CNT 61 THERE ARE 61 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L7 ANSWER 4 OF 11 MEDLINE DUPLICATE 1
AN 2002126522 MEDLINE
DN 21851318 PubMed ID: 11861924
TI Single nucleotide polymorphism detection by combinatorial fluorescence energy transfer tags and biotinylated dideoxynucleotides.
AU Tong Anthony K; Ju Jingyue
CS Laboratory of DNA Sequencing and Chemical Biology, Columbia Genome Center, Columbia University College of Physicians and Surgeons, 1150 St Nicholas Avenue, New York, NY 10032, USA.
SO NUCLEIC ACIDS RESEARCH, (2002 Mar 1) 30 (5) e19.
Journal code: 0411011. ISSN: 1362-4962.
CY England: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 200203
ED Entered STN: 20020226
Last Updated on STN: 20020312
Entered Medline: 20020311
AB Combinatorial fluorescence energy transfer (CFET) tags, constructed by exploiting energy transfer and combinatorial synthesis, allow multiple biological targets to be analyzed simultaneously. We here describe a multiplex single nucleotide polymorphism (SNP) assay based on **single base extension** (SBE) using CFET tags and biotinylated dideoxynucleotides (biotin-ddNTPs). A library of CFET-labeled oligonucleotide primers was mixed with biotin-ddNTPs, DNA polymerase and the DNA templates containing the SNPs in a single tube. The nucleotide at the 3'-end of each CFET-labeled oligonucleotide primer was complementary to a particular SNP in the template. Only the CFET-labeled primer that is fully complementary to the DNA template was extended by DNA polymerase with a biotin-ddNTP. We isolated the DNA extension fragments that carry a biotin at the 3'-end by capture with streptavidin-coated magnetic beads, while the unextended primers were eliminated. The biotinylated fluorescent DNA fragments were subsequently analyzed in a multicolor fluorescence electrophoresis system. The distinct fluorescence signature and electrophoretic mobility of each DNA extension product in the electropherogram coded the SNPs without the use of a sizing standard. We simultaneously distinguished six nucleotide variations in synthetic DNA templates and a PCR product from the retinoblastoma tumor suppressor gene. The use of CFET-labeled primers and biotin-ddNTPs **coupled** with the specificity of DNA polymerase in SBE offered a multiplex method for detecting SNPs.

L7 ANSWER 5 OF 11 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2002:233363 BIOSIS
DN PREV200200233363
TI Single tube SNP genotyping using mini-extension **coupled** with IR-labeled AcycloTerminatorsTM.
AU Kovar, J. (1); Qiu, J. (1); Olive, M. (1)
CS (1) LI-COR, Inc., Lincoln, NE USA
SO Abstracts of the General Meeting of the American Society for Microbiology, (2001) Vol. 101, pp. 734. http://www.asmusa.org/mtgsrc/generalmeeting.htm. print.
Meeting Info.: 101st General Meeting of the American Society for Microbiology Orlando, FL, USA May 20-24, 2001
ISSN: 1060-2011.
DT Conference
LA English
AB The popularity of single nucleotide polymorphism (SNP) detection has led to the rapid development of several methodologies. The use of SNPs, however, is still limited due to the difficulty of linking SNPs (especially those non-coding SNPs) to a target phenotype(s). In the

present study, we illustrate how a single base change between the normal (wild-type) and mutant genes in Chlamydomonas reinhardtii (green algae) acetolactate synthase (ALS) gene can be detected using a mini-extension method **coupled** with a newly developed IR-labeled AcycloTerminatorTM (NEN Life Technologies, Boston, MA). A clone containing the ALS gene was isolated from a cDNA library and sequenced. Site-directed mutagenesis was used to incorporate single base changes within the wild-type gene that correspond with known differences between wild-type and mutant strains of Chlamydomonas ALS. Amplification of specific regions of interest was done using PCR to narrow the region of analysis. Mini-Extension involved the use of a primer adjacent to the polymorphic site and a mixture containing one AcycloTerminator (corresponding to either the wild-type or mutant base for the known polymorphism) with the three remaining dNTPs. The one tube assay yielded two possible products, one when the AcycloTerminator (corresponding to the mutant base present) was incorporated at the polymorphism site and the other when the AcycloTerminator was incorporated downstream (corresponding to the wild-type). When compared with **single base extension** (four tube assay), the mini-extension based SNP procedure offers a high-throughput solution. A detailed protocol will be presented.

L7 ANSWER 6 OF 11 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2002:70245 BIOSIS
DN PREV200200070245
TI **Coupled** analysis of DHPLC and **single-base extension** to localize and genotype mutations in the HFE gene.
AU Marino, M. A. (1); McAndrew, P. E. (1); Sharma, A. (1); Woolcock, C. (1); Devaney, J. M.
CS (1) Applied Genomics and Molecular Genetics, Transgenomic Inc, Gaithersburg, MD USA
SO American Journal of Human Genetics, (October, 2001) Vol. 69, No. 4 Supplement, pp. 633. http://www.journals.uchicago.edu/AJHG/home.html. print.
Meeting Info.: 51st Annual Meeting of the American Society of Human Genetics San Diego, California, USA October 12-16, 2001
ISSN: 0002-9297.
DT Conference
LA English

L7 ANSWER 7 OF 11 MEDLINE DUPLICATE 2
AN 2001142347 MEDLINE
DN 21085587 PubMed ID: 11217771
TI Genotyping of two mutations in the HFE gene using **single-base extension** and high-performance liquid chromatography.
AU Devaney J M; Pettit E L; Kaler S G; Vallone P M; Butler J M; Marino M A
CS Transgenomic Inc., Gaithersburg, Maryland 20878, USA.. jdevaney@transgenomic.com
SO ANALYTICAL CHEMISTRY, (2001 Feb 1) 73 (3) 620-4.
Journal code: 0370536. ISSN: 0003-2700.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 200103
ED Entered STN: 20010404
Last Updated on STN: 20010404
Entered Medline: 20010308
AB Currently, a major focus of human genetics is the utilization of single-nucleotide polymorphisms for clinical diagnostics, whole-genome linkage disequilibrium screens to identify common disease genes such as

Alzheimer disease, determination of the recent evolutionary history of a species, and the process of speciation. We have examined **single-nucleotide extension coupled** with high-performance liquid chromatography as a method to simultaneously genotype two SNPs occurring in the coding region of the HFE gene that produce clinical effects. This assay allows concurrent genotyping of the C282Y and H63D mutations in 11 min and is 100% concordant with current testing methods for both of these mutations.

L7 ANSWER 8 OF 11 MEDLINE DUPLICATE 3
AN 2001156126 MEDLINE
DN 21098045 PubMed ID: 11159763
TI Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies.
AU Shi M M
CS Department of Applied Genomics, Genometrix Inc., The Woodlands, TX 77381, USA.. mshi@genometrix.com
SO CLINICAL CHEMISTRY, (2001 Feb) 47 (2) 164-72. Ref: 40
Journal code: 9421549. ISSN: 0009-9147.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LA English
FS Priority Journals
EM 200103
ED Entered STN: 20010404
Last Updated on STN: 20010404
Entered Medline: 20010322
AB BACKGROUND: Pharmacogenetics is a scientific discipline that examines the genetic basis for individual variations in response to therapeutics. Pharmacogenetics promises to develop individualized medicines tailored to patients' genotypes. However, identifying and genotyping a vast number of genetic polymorphisms in large populations also pose a great challenge. APPROACH: This article reviews the recent technology development in mutation detection and genotyping with a focus on genotyping of single nucleotide polymorphisms (SNPs). CONTENT: Novel mutations/polymorphisms are commonly identified by conformation-based mutation screening and direct high-throughput heterozygote sequencing. With a large amount of public sequence information available, in silico SNP mapping has also emerged as a cost-efficient way for new polymorphism identification. Gel electrophoresis-based genotyping methods for known polymorphisms include PCR **coupled** with restriction fragment length polymorphism analysis, multiplex PCR, oligonucleotide ligation assay, and minisequencing. Fluorescent dye-based genotyping technologies are emerging as high-throughput genotyping platforms, including oligonucleotide ligation assay, pyrosequencing, **single-base extension** with fluorescence detection, homogeneous solution hybridization such as TaqMan, and molecular beacon genotyping. Rolling circle amplification and Invader assays are able to genotype directly from genomic DNA without PCR amplification. DNA chip-based microarray and mass spectrometry genotyping technologies are the latest development in the genotyping arena. SUMMARY: Large-scale genotyping is crucial to the identification of the genetic make-ups that underlie the onset of diseases and individual variations in drug responses. Enabling technologies to identify genetic polymorphisms rapidly, accurately, and cost effectively will dramatically impact future drug and development processes.

L7 ANSWER 9 OF 11 CAPLUS COPYRIGHT 2002 ACS
AN 2000:772797 CAPLUS
DN 133:345529
TI Primer extension on a microarray of gel-immobilized primers

IN Dubiley, Svetlana; Kirillov, Eugene; Mirzabekov, Andrei
PA University of Chicago, USA
SO PCT Int. Appl., 35 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2000065098	A2	20001102	WO 2000-US11286	20000425
	WO 2000065098	A3	20010719		
	W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CR, CU, CZ, DE, DK, DM, DZ, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM				
	RW: GH, GM, KE, LS, MW, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG				
	EP 1171637	A2	20020116	EP 2000-928451	20000425
	R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT, IE, SI, LT, LV, FI, RO				
PRAI	US 1999-300675	A	19990427		
	WO 2000-US11286	W	20000425		

AB Methods and compns. have been developed for nucleotide extension of primers immobilized within gel pads on a microchip using multibase primers or multiple sets of primers, or combinations thereof. Mols. or parts of mols. are identified. The effect of the different temp., reaction time are tested. The **single base extension** was amplified by carrying out the reaction under elevated temp. The invention is exemplified by detecting B. anthracis toxin gene (pag or lef) , diagnosing seven commonly occurring .beta.-thalassemia mutations within .beta.-globin gene, and detecting a specific antibody in a library of antibodies by **coupling** each antibody with labeled nucleic acid tags. The method is useful to detect single nucleotide mutations for genetic diagnosis, and specific antibody to a particular antigen.

L7 ANSWER 10 OF 11 CAPLUS COPYRIGHT 2002 ACS
AN 2000:707335 CAPLUS
DN 133:291910
TI Ordered addressable arrays of oligonucleotides for use as a general substrate in the preparation of probe arrays
IN Fan, Jian-Bing; Hirschhorn, Joel N.; Huang, Xiaohua; Kaplan, Paul; Lander, Eric S.; Lockhart, David J.; Ryder, Thomas; Sklar, Pamela
PA Whitehead Institute for Biomedical Research, USA; Affymetrix, Inc.
SO PCT Int. Appl., 83 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2000058516	A2	20001005	WO 2000-US8069	20000327
	WO 2000058516	A3	20010719		
	W: CA, JP				
	RW: AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE				
	EP 1165839	A2	20020102	EP 2000-918432	20000327
	R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT, IE, FI				
PRAI	US 1999-126473P	P	19990326		

US 1999-140359P P 19990623
WO 2000-US8069 W 20000327

AB An array of oligonucleotides on a solid substrate is disclosed, which can be used for multiple purposes. Oligonucleotides at one site on the array have the same distinct sequence that can be use to capture a probe carrying the complementary sequence. Libraries of probes with the appropriate sequences can assemble themselves on the immobilized array and can be removed as needed for the prepn. of a new array on the surface. Methods and reagents are provided for performing genotyping to det. the identity or ration of allelic forms of a gene in a sample. A **single base extension** primer is **coupled** to a sequence identity code. During the primer extension reaction a distinctive label is incorporated which identifies the allelic form present in the sample. This permits multiple simultaneous analyses to be performed easily and efficiently. Use of the method to identify alleles of a gene and of rare sequences, e.g. arising from somatic mutation, are demonstrated.

L7 ANSWER 11 OF 11 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 2001:151558 BIOSIS
DN PREV200100151558
TI Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies.
AU Shi, Michael M. (1)
CS (1) Genometrix Inc., 2700 Research Forest Dr., The Woodlands, TX, 77381: mshi@genometrix.com USA
SO Clinical Chemistry, (February, 2000) Vol. 47, No. 2, pp. 164-172. print. ISSN: 0009-9147.
DT General Review
LA English
SL English
AB Background: Pharmacogenetics is a scientific discipline that examines the genetic basis for individual variations in response to therapeutics. Pharmacogenetics promises to develop individualized medicines tailored to patients' genotypes. However, identifying and genotyping a vast number of genetic polymorphisms in large populations also pose a great challenge. Approach: This article reviews the recent technology development in mutation detection and genotyping with a focus on genotyping of single nucleotide polymorphisms (SNPs). Content: Novel mutations/polymorphisms are commonly identified by conformation-based mutation screening and direct high-throughput heterozygote sequencing. With a large amount of public sequence information available, in silico SNP mapping has also emerged as a cost-efficient way for new polymorphism identification. Gel electrophoresis-based genotyping methods for known polymorphisms include PCR **coupled** with restriction fragment length polymorphism analysis, multiplex PCR, oligonucleotide ligation assay, and minisequencing. Fluorescent dye-based genotyping technologies are emerging as high-throughput genotyping platforms, including oligonucleotide ligation assay, pyrosequencing, **single-base extension** with fluorescence detection, homogeneous solution hybridization such as TaqMan(R), and molecular beacon genotyping. Rolling circle amplification and InvaderTM assays are able to genotype directly from genomic DNA without PCR amplification. DNA chip-based microarray and mass spectrometry genotyping technologies are the latest development in the genotyping arena. Summary: Large-scale genotyping is crucial to the identification of the genetic make-ups that underlie the onset of diseases and individual variations in drug responses. Enabling technologies to identify genetic polymorphisms rapidly, accurately, and cost effectively will dramatically impact future drug and development processes.

=> d his

(FILE 'HOME' ENTERED AT 11:20:22 ON 22 JUL 2002)

FILE 'MEDLINE, BIOSIS, CAPLUS' ENTERED AT 11:26:44 ON 22 JUL 2002
L1 147 S SINGLE (W) (NUCLEOTIDE OR BASE) (W) EXTEN?
L2 3 S L1 AND (ROLLING (W) CIRCLE)
L3 0 S L1 AND LIZARDI?/AU
L4 48 S L1 AND AMPLIF?
L5 32 DUP REM L4 (16 DUPLICATES REMOVED)
L6 14 S L1 AND COUPL?
L7 11 DUP REM L6 (3 DUPLICATES REMOVED)

=> s rolling (w)circle or rca
L8 7357 ROLLING (W) CIRCLE OR RCA

=> s l8 and l1
L9 3 L8 AND L1

=> d 1 bib

L9 ANSWER 1 OF 3 MEDLINE
AN 2001156126 MEDLINE
DN 21098045 PubMed ID: 11159763
TI Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies.
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